

# Ange-Line Bruel

## List of Publications by Year in descending order

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Version: 2024-02-01

48  
papers

1,520  
citations

471509

17  
h-index

345221

36  
g-index

48  
all docs

48  
docs citations

48  
times ranked

3719  
citing authors

#	ARTICLE	IF	CITATIONS
1	High efficiency and clinical relevance of exome sequencing in the daily practice of neurogenetics. <i>Journal of Medical Genetics</i> , 2022, 59, 445-452.	3.2	6
2	ITSN1: a novel candidate gene involved in autosomal dominant neurodevelopmental disorder spectrum. <i>European Journal of Human Genetics</i> , 2022, 30, 111-116.	2.8	4
3	MYT1L-associated neurodevelopmental disorder: description of 40 new cases and literature review of clinical and molecular aspects. <i>Human Genetics</i> , 2022, 141, 65-80.	3.8	14
4	Accelerated genome sequencing with controlled costs for infants in intensive care units: a feasibility study in a French hospital network. <i>European Journal of Human Genetics</i> , 2022, 30, 567-576.	2.8	12
5	Copy number variants calling from WES data through eXome hidden Markov model (XHMM) identifies additional 2.5% pathogenic genomic imbalances smaller than 30Åkb undetected by arrayâ€CGH. <i>Annals of Human Genetics</i> , 2022, 86, 171-180.	0.8	6
6	Expanding the phenotype of <sc><i>HNRNPU</i></sc>-related neurodevelopmental disorder with emphasis on seizure phenotype and review of literature. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1497-1514.	1.2	6
7	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. <i>American Journal of Human Genetics</i> , 2022, 109, 601-617.	6.2	16
8	Toward clinical and molecular dissection of frontonasal dysplasia with facial skin polyps: From Pai syndrome to differential diagnosis through a series of 27 patients. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2036-2047.	1.2	1
9	Atypical phenotype of a patient with Bardetâ€Biedl syndrome type 4. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2022, 10, e1869.	1.2	2
10	Same performance of exome sequencing before and after fetal autopsy for congenital abnormalities: toward a paradigm shift in prenatal diagnosis?. <i>European Journal of Human Genetics</i> , 2022, , .	2.8	1
11	Genotype-first in a cohort of 95 fetuses with multiple congenital abnormalities: when exome sequencing reveals unexpected fetal phenotype-genotype correlations. <i>Journal of Medical Genetics</i> , 2021, 58, 400-413.	3.2	18
12	Neuropsychological study in 19 French patients with <sc>Whiteâ€Sutton</sc> syndrome and <sc><i>POGZ</i></sc> mutations. <i>Clinical Genetics</i> , 2021, 99, 407-417.	2.0	10
13	DLG4-related synaptopathy: a new rare brain disorder. <i>Genetics in Medicine</i> , 2021, 23, 888-899.	2.4	16
14	<sc>Skraabanâ€Deardorff</sc> syndrome: Six new cases of <sc><i>WDR</i>26</sc>-related disease and expansion of the clinical phenotype. <i>Clinical Genetics</i> , 2021, 99, 732-739.	2.0	4
15	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. <i>American Journal of Human Genetics</i> , 2021, 108, 346-356.	6.2	30
16	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. <i>Human Mutation</i> , 2021, 42, 445-459.	2.5	26
17	Lossâ€ofâ€function variants in ARHGEF9 are associated with an Xâ€linked intellectual disability dominant disorder. <i>Human Mutation</i> , 2021, 42, 498-505.	2.5	1
18	Expanding the phenotype of <sc><i>ASXL3</i></sc>-related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <sc><i>ASXL3</i></sc>. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3446-3458.	1.2	12

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19	The diagnostic rate of inherited metabolic disorders by exome sequencing in a cohort of 547 individuals with developmental disorders. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 29, 100812.	1.1	2
20	Interest of exome sequencing trio-like strategy based on pooled parental DNA for diagnosis and translational research in rare diseases. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2021, 9, e1836.	1.2	5
21	De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaides-Baraitser syndrome. <i>Genetics in Medicine</i> , 2020, 22, 1838-1850.	2.4	31
22	Hydrothorax in fetal cases of Opitz G/BBB diagnosis: Extending the phenotype?. <i>Clinical Genetics</i> , 2020, 98, 620-621.	2.0	1
23	Second-tier trio exome sequencing after negative solo clinical exome sequencing: an efficient strategy to increase diagnostic yield and decipher molecular bases in undiagnosed developmental disorders. <i>Human Genetics</i> , 2020, 139, 1381-1390.	3.8	8
24	Excess of de novo variants in genes involved in chromatin remodelling in patients with marfanoid habitus and intellectual disability. <i>Journal of Medical Genetics</i> , 2020, 57, 466-474.	3.2	7
25	Next-generation sequencing approaches and challenges in the diagnosis of developmental anomalies and intellectual disability. <i>Clinical Genetics</i> , 2020, 98, 433-444.	2.0	20
26	Kosaki overgrowth syndrome: A novel pathogenic variant in PDGFRB and expansion of the phenotype including cerebrovascular complications. <i>Clinical Genetics</i> , 2020, 98, 19-31.	2.0	17
27	Deciphering exome sequencing data: Bringing mitochondrial DNA variants to light. <i>Human Mutation</i> , 2019, 40, 2430-2443.	2.5	11
28	Increased diagnostic and new genes identification outcome using research reanalysis of singleton exome sequencing. <i>European Journal of Human Genetics</i> , 2019, 27, 1519-1531.	2.8	43
29	Variant recurrence in neurodevelopmental disorders: the use of publicly available genomic data identifies clinically relevant pathogenic missense variants. <i>Genetics in Medicine</i> , 2019, 21, 2504-2511.	2.4	21
30	2.5 years' experience of GeneMatcher data-sharing: a powerful tool for identifying new genes responsible for rare diseases. <i>Genetics in Medicine</i> , 2019, 21, 1657-1661.	2.4	14
31	INTU-related oral-facial-digital syndrome type VI: A confirmatory report. <i>Clinical Genetics</i> , 2018, 93, 1205-1209.	2.0	7
32	Unexpected diagnosis of a SHH nonsense variant causing a variable phenotype ranging from familial coloboma and Intellectual disability to isolated microcephaly. <i>Clinical Genetics</i> , 2018, 94, 182-184.	2.0	2
33	Truncating variants of the DLG4 gene are responsible for intellectual disability with marfanoid features. <i>Clinical Genetics</i> , 2018, 93, 1172-1178.	2.0	19
34	A Recurrent De Novo PACS2 Heterozygous Missense Variant Causes Neonatal-Onset Developmental Epileptic Encephalopathy, Facial Dysmorphism, and Cerebellar Dysgenesis. <i>American Journal of Human Genetics</i> , 2018, 102, 995-1007.	6.2	49
35	Loss-of-Function Mutations in UNC45A Cause a Syndrome Associating Cholestasis, Diarrhea, Impaired Hearing, and Bone Fragility. <i>American Journal of Human Genetics</i> , 2018, 102, 364-374.	6.2	40
36	Clinical whole-exome sequencing for the diagnosis of rare disorders with congenital anomalies and/or intellectual disability: substantial interest of prospective annual reanalysis. <i>Genetics in Medicine</i> , 2018, 20, 645-654.	2.4	146

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37	Okurâ€Chung neurodevelopmental syndrome: Eight additional cases with implications on phenotype and genotype expansion. <i>Clinical Genetics</i> , 2018, 93, 880-890.	2.0	30
38	The oculoauriculofrontonasal syndrome: Further clinical characterization and additional evidence suggesting a nontraditional mode of inheritance. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2740-2750.	1.2	6
39	De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. <i>Nature Genetics</i> , 2018, 50, 1442-1451.	21.4	28
40	Autosomal recessive truncating <i>MAB21L1</i> mutation associated with a syndromic scrotal agenesis. <i>Clinical Genetics</i> , 2017, 91, 333-338.	2.0	15
41	Fifteen years of research on oralâ€facialâ€digital syndromes: from 1 to 16 causal genes. <i>Journal of Medical Genetics</i> , 2017, 54, 371-380.	3.2	85
42	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2017, 101, 664-685.	6.2	337
43	Expanding the clinical spectrum of recessive truncating mutations of <i>KLHL7</i> to a Bohring-Opitz-like phenotype. <i>Journal of Medical Genetics</i> , 2017, 54, 830-835.	3.2	15
44	Reducing diagnostic turnaround times of exome sequencing for families requiring timely diagnoses. <i>European Journal of Medical Genetics</i> , 2017, 60, 595-604.	1.3	22
45	Dominant variants in the splicing factor <i>PUF60</i> cause a recognizable syndrome with intellectual disability, heart defects and short stature. <i>European Journal of Human Genetics</i> , 2017, 25, 43-51.	2.8	44
46	The ciliopathy-associated <i>CPLANE</i> proteins direct basal body recruitment of intraflagellar transport machinery. <i>Nature Genetics</i> , 2016, 48, 648-656.	21.4	119
47	Expanding the Phenotype Associated with <i>NAA10</i> -Related N-Terminal Acetylation Deficiency. <i>Human Mutation</i> , 2016, 37, 755-764.	2.5	70
48	The oral-facial-digital syndrome gene <i>C2CD3</i> encodes a positive regulator of centriole elongation. <i>Nature Genetics</i> , 2014, 46, 905-911.	21.4	121